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COGNITIVE AND AFFECTIVE IMPAIRMENTS OF A NOVEL SCA/MND CROSSROAD MUTATION ASIDAN

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Background: A variety of hereditary spinocerebellar ataxia (SCA) develops a broad spectrum of both ataxia and non-ataxia symptoms. Cognitive and affective changes are one such non-ataxia symptoms, but have been described only in hereditary SCAs with exonic CAG gene expansion.
Methods: We newly found intronic hexanucleotide GGCCTG gene expansion in NOP56 gene as the causative mutation (=SCA36) in nine unrelated Japanese familial SCA originating from Asida river area in the western part of Japan, thus nicknamed Asidan for this mutation. These patients show unique clinical balance of cerebellar ataxia and motor neuron disease (MND), locating on the crossroad of these two diseases. We examined cognitive and affective analyses on 12 Asidan patients who agreed to join the examination.
Results: The 12 Asidan patients demonstrated a significant decrease in their frontal executive functions measured by frontal assessment battery (FAB) and Montreal cognitive assessment (MoCA) compared with age- and gender-matched controls, whilst mini-mental state examination (MMSE) and Hasegawa dementia score-revised (HDS-R) were within normal range. The decline of frontal executive function was related to their disease duration and scale for the assessment and rating of ataxias (SARA). They also demonstrated mild depression and apathy. Single-photon emission tomography (SPECT) analysis showed that these Asidan patients showed decline of regional cerebral blood flow (rCBF) in a particular areas of cerebral cortices such as Brodmann areas 24 and 44-46.
Conclusions: These data suggest the patients with Asidan mutation show unique cognitive and affective characteristics different from other hereditary SCAs with exonal CAG expansion or MND.

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